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ABSTRACT

This paper presents a case study of Gorlin Syndrome, also known as Basal Cell Nevus Syndrome, a rare genetic disorder characterized by widespread developmental defects. Criteria for diagnosis are listed, noting the presence of frequent basal cell carcinomas at a relatively young age and multiple cysts of the jaw. Speech and/or language impairments may result from malocclusion, possible cleft palate, and frequent surgeries. The case study is of a 19-year-old female college student who already had 10 surgeries for malignant jaw cysts. Results of hearing, speech, and language tests found normal hearing, expressive and receptive language skills within normal limits, and articulation within normal limits. However, the Dworkin-Culatta Oral Mechanism Examination found numerous abnormalities and weaknesses. Recommendations for speech therapy services to strengthen musculature on one side were made. Decreased muscle flaccidity were noted following the intervention. (DB)

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Manifestations, Treatment Implications and Speech and Language Considerations of Gorlin Syndrome: A Case Study

Introduction

Gorlin Syndrome, also known as Basal Cell Nevus Syndrome, is rare with 600 identified cases. It is an autosomal dominant disorder mapped to chromosome 9q22-q31, and allelic loss at this location is common in tumors from Gorlin Syndrome patients (Shimkets, et.al., 1996) This syndrome features widespread developmental defects (Gailani, et.al. 1992).

There are about 100 features of Gorlin Syndrome Major features include basal cell carcinomas, which manifest in cysts of the jaw and skeleton. Odontogenic keratocysts or cysts of the jaw occur in the 1st decade of life, usually about the 7th year and peak in the 2nd and 3rd decades of life (Gorlin, 1987). These cysts are found about 3 times as often in the mandible as in the maxilla. Characteristics facies are present in about 70% of cases and include frontal bulging (Gorlin, 1987). Other features include macrocephaly, rib abnormalities, scoliosis, strabismus and occasional cognitive impairment (Shprintzen, 1997; Gorlin, 1987). Cleft palates occur in about 5% of the cases, and a compensatory articulation disorder may occur if the patient has a cleft palate. Hypernasality may be found secondary to the cleft in a small percentage of patients (Shprintzen, 1997). Hearing acuity and voice are normal,, but language may be delayed or impaired in some cases with central nervous system anomalies and cognitive impairment. Additional features include medulloblastomas, ovarian fibromas, mild hypertelorism, exaggerated length of the mandible and well developed supraorbital

ridges giving the eyes a sunken appearance. Gorlin (1987) reported that there may be fused eyebrows, a broadened nasal root and pouting of the lower lip associated with the exaggerated length of the mandible.

Diagnosis requires 2 major or 1 major and 1 minor criteria. Major criteria includes: (1) more than 2 basal cell carcinomas; (2) less that 30 years of age, cysts of the jaw (odotogenic keratocyst, palmar pits, falx calcification and family history. Minor criteria include: rib or vertebral anomalies, macrocrania, fibromas, medulloblastomas and lymphomesenteric cysts (Gerhardt, 1996).

Of interest to the speech-language pathologist may be the characteristics of a Class III malocclusion (prognathic jaw), high vaulted palate, lateral open bite and possible palatal cleft, possible cognitive impairments, possible language delays or language impairments and the characteristic facial structure. These characteristics may affect speech and/or language production and feeding.

Case Study

A 19-year-old female college student with Gorlin Syndrome was seen at a university speech and hearing clinic. Referral was made by her plastic surgeon from a medical college because of left-sided facial flaccidity. History revealed that the client was diagnosed with Gorlin Syndrome, also known as Basal Cell Nevus Syndrome, at the age of 1 1/2 . The client's father also has this syndrome.

Because of numerous malignant jaw cysts, there have been 10 surgeries as follows:

1981: resection of 2 mandibular cysts

1983: resection of 1 cyst

1985: resection of 1 cyst

1987: resection of 2 maxillary cysts

1988: resection of 1 cyst

1990: resection of 1 cyst

1992: resection of 2 mandibular cysts and ectopic tooth, left maxillary sinus, with iliac bone graft to mandible

1996: resection of 2 cysts, 1 in left maxillary sinus

1998: 2 cysts removed from ovaries

1998: jaw reconstructive surgery

Because of the cysts and surgeries, the client reported that she had abnormal growth of the lower jaw, a Class III occlusal relationship, weak projection of the cheekbones, a large v-shaped deformity at the front of the lower jaw and teeth contact only in the back, giving her an open bite. She reported that she has had severe dental crowding and facial proportions described by her physician as upper midface hypoplasia, malar hypoplasia, an elongated nose and central philtral area with a long midface.

Even though the client was a college freshman, with average to above average grades, a graduate level clinician administered a complete battery of speech and language testing because of reported language and speech problems in clients with Gorlin Syndrome. This clinician was supervised by an Assistant Professor in Speech/Language Pathology who had completed the Deep Pharyngeal Neuromuscular Stimulation training.

Hearing:

A pure tone-hearing test was administered at 25 dB bilaterally. The client passed this test.

Language:

The Test of Adolescent and Adult Language – 3 (TOAL 3) was administered to assess receptive and expressive language abilities.

Listening Vocabulary	50th percentile
Listening Grammar	63rd percentile
Speaking Vocabulary	25th percentile
Speaking Grammar	63rd percentile
Reading Vocabulary	25th percentile
Reading Grammar	63rd percentile
Writing Vocabulary	75th percentile
Writing Grammar	63rd percentile

All of the following scores fell with the average range for her age. Thus it was concluded that her expressive and receptive language skills were within normal limits.

Articulation

The reading portion of the Test of Minimal Articulation Competence (T-MAC) was administered to assess articulation skills. The client obtained a total score of 117 with an articulation index of very mild. The only articulation error noted was a slight distortion of /z/ in the medial position of words. This error was somewhat visually distracting; however, it did not influence her intelligibility. Thus, her articulation was rated within normal limits.

Oral Mechanism Exam

The Dworkin-Culatta Oral Mechanism Examination was administered to examine the oral structures and their function. The following disabilities were noted:

- Left and right jaw and chin area sensation diminished
- Range of lip protrusion reduced – deviation to left
- Imprecise movement of lips – left side appeared weaker
- High narrow arch
- Tongue mildly deviated to left upon protrusion
- Reduced strength of tongue on left side
- Flaccidity of tongue on left side
- Diadochokinetic rate – abnormal syllable stress pattern
- Open bite – left lateral

The client further noted that because of the above-mentioned weaknesses she sometimes had trouble with eating ,especially with biting and then tearing sandwiches. However, she had compensated for these weaknesses by the foods that she chose and slowing down her pace of eating.

Voice and Fluency

Both of these areas were judged to be within normal limits for her age and gender in conversational speech.

Clinical Impressions

The client was found to have muscle flaccidity on the entire left side of her face, including her tongue. Her right side of her face was found to be much stronger than the left.

Prognosis

Stimulability for muscle strengthening of the facial area was judged to be a strength; thus, this intervention was considered appropriate for this client. Because of her self-motivation and cognitive awareness of oral-motor deficits, it was concluded that her prognosis for change was excellent.

Recommendations:

It is recommended that this client receive speech therapy services for one-hour sessions twice weekly. Treatment should focus on:

- Oral-motor excerises specific to the areas that showed flaccidity during the evaluation of the left side of the face
- Icing and massage to strengthen the left side of the face to obtain facial and oral symmetry
- A home program of oral motor exercises twice a day

Intervention:

This client was seen twice a week for one hour at the speech and hearing clinic. A system of icing, massage, and active exercise was initiated. Electrical stimulation (E-stim) therapy was considered as possible treatment. It was contraindicated because of titanium rods placed in the jaw as a result of surgeries.

The client's left facial muscles levator lavii aleque nase, levator labii, zygomatic minor, zygomatic major, obicularis oris, masseter, depressor anguli oris, depressor labii inferioris, risorius/buccinator and mentalis were iced. This procedure involved the application of ice over the target muscle by applying firm, even pressure along the muscle in the direction of the muscle movement. The ice was applied for 3-10 seconds.

This was followed by massage. The clinician applied firm even pressure along the muscle in the direction of the muscle movement.

Finally, oral motor exercises that included the major muscle groups of the face were targeted. The client was directed to imitate the oral movements of the clinician. A large mirror was used to facilitate visual feedback.

After all major muscle groups were iced, massaged and exercised another round of stimulation was performed. Usually each muscle received three episodes of stimulation with a brief rest period between each episode of stimulation.

Results

Muscles of the left side of the face were considered less flaccid. Left side of the mouth was not as turned down. Smile was more symmetrical. At rest, the lips were symmetrical. When at rest, the muscles of cheek and mouth were more taut and judged by the graduate clinician and the client to have more strength. Overall, this intervention was viewed as successful for this client.

Note

This paper describes a single case study on a college student who had been diagnosed with Gorlin Syndrome at the age of 1 ½. Her manifestations of this syndrome may be different from others diagnosed with the same syndrome. The approach described in this paper was specific to this client and may not be appropriate for all clients with Gorlin Syndrome

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